CLINICAL SPECTRUM OF LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 2E (LGMD2E)

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Autosomal recessive muscular dystrophies are commonly seen. Although classification systems have changed over the years, the latest classification of muscular dystrophies is according to the type of genetic mutation and clinical features.

Limb-girdle muscular dystrophy (LGMD) 2E is caused by a mutation in the beta sarcoglycan gene. Proximal muscle weakness in the shoulder and pelvic girdle muscles is the main clinical feature. Facial muscle weakness is mild or absent. Muscle wasting occurs in early childhood or adulthood. The involvement of the pelvic muscles is more prominent than shoulder muscle involvement in those with childhood-onset. Eye muscles are spared. Ambulation is lost at different stages of life, depending on the course of the disease. Cardiac and respiratory complications start to emerge, mostly when the patient becomes nonambulatory.

OBJECTIVES

To contribute to the literature, we collect and present the demographic information and clinical data of patients diagnosed with LGMD2E in Turkey.

RESULTS

We evaluated nine patients previously diagnosed with LGMD2E. 4 children were female, 5 children were male and 3 siblings have included in the study. Except for 2 of the patients, all of them were diagnosed around the age of 10. At the time of diagnosis, the patient's complaints were fatigue, difficulty in walking and climbing stairs. Incidental elevation of LFT was detected in 2 patients' blood samples and after performing further examination they were diagnosed as LGMDE2. All patients have consanguineous parents.

In their physical examination, 3 of 9 patients had muscle hypertrophy, while 4 patients had muscle atrophy. Facial myopathy was present in 4 patients. 7 patients had generalized muscle weakness. Only 2 children had wing scapula and 6 children had joint contractures especially in the knee and ankle joints. 3 children were ambulatory but Gowers' signs were prolonged for 3 and 7 seconds. Mutations detected in patients' genetic tests are c.686A>T, c.1A>T and c.610T>C.

	gender	age of diagnosis	muscle atrophy	muscle hypertrophy	facial myopathy	wing scapula	contractures	ambulation	cardiac involvement
Patient 1	Female	9y	+	-	+	-	+	NA	+
Patient 2	Female	7y	+	-	+	+	+	NA	+
Patient 3	Male	10y	-	-	-	-	-	A	-
Patient 4	Female	8y	-	+	-	-	+	NA	-
Patient 5	Male	10y	-	+	-	-	+	NA	-
Patient 6	Male	9y	+	-	+	+	+	NA	+
Patient 7	Male	11y	-	+	-	-	-	A	-
Patient 8	Female	10y	-	-	-	-	-	A	-
Patient 9	male	7y	+	-	+	-	+	NA	-

CONCLUSION

LGMD2E can present a DMD-like course with onset in the first decade of life. There may develop cardiorespiratory involvement. In LGMD2E, gene therapy efforts are ongoing for the past few years. This is why our cohort is actually a natural history study. With natural history studies and advances in molecular medicine, our understanding of the pathophysiology and disease progression are augmenting.

REFERENCES

- Vainzof M, Souza LS, Gurgel-Giannetti J, Zatz M. Sarcoglycanopathies: an update. *Neuromuscul Disord*. 2021;31(10):1021-1027.
- Semplicini C, Vissing J, Dahlqvist JR, et al. Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. *Neurology*. 2015;84(17):1772-1781